

REPLACEMENT SHEET

Figure 1

MAMSSGGSGGVPEQEDSVLFRRGTQSDDSDIWDDTALIKAYDAVASFKHALKNG  
DICETSGKPCTPKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPATIA  
SIDFKRETCVVVTGYGNREEQNLSDLSPICEVANNIEQNAQENENESQVSTDESE  
NSRSPGNKSDNIKPKSAPWNSFLPPPPMPGPRLGPGKPGLKFNNGPPPPPPPHL  
LSCWLPPFPGPPIIPPPPICPDSSLDDADALGSMLISWYMSGYHTGYYMGFRQNQK  
EGRCSHSLN



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**REPLACEMENT SHEET**

**Figure 2A**

REPLACEMENT SHEET

Figure 2B

AATTTTAAATTTGTAGAGACAGGGTCTCATTATGTTGCCAGGGTGGTCAA  
GCTCCAGGTCTCAAGTGATCCCCCTACCTCCGCCTCCAAAGTTGTGGGATTGTAGG  
CATGAGCCACTGCAAGAAAACCTTAAC TG CAG CCT AATA ATT GT TT CTT GGG ATA  
ACTTTAAAGTACATTAAAAGACTATCAACTTAATTCTGATCATATTGTGAAT  
AAAATAAGTAAAATGTCTGTGAACAAAATGCTTTAACATCCATATAAAGCTATC  
TATATATAGCTATCTATCTATAGCTATTTTTAACCTCCTTTATTCCT  
TACAG\*GGTTTAGACAAAATCAAAAAGAAGGAAGGTGCTCACATTCTAAATTAA  
GGA\*GTAAGTCTGCCAGCATTATGAAAGTGAATCTTACTTTGTAAAACCTTATGGT  
TTGTGGAAAACAAATGTTTGAAACAGTTAAAAGTTAGATGTTAGAAAGTTGAAA  
GGTTAATGTAAAACAATCAATATTAAAGAATTGATGCCAAAACATTAGATAAAA  
GGTTAATCTACATCCCTACTAGAATTCTCATACTTAACGGTTGGTTGTGGAAGA  
AACATACTTCACAATAAAGAGCTTTAGGATATGATGCCATTATCACTAGTAG  
GCAGACCAGCAGACTTTTTATTGTGATATGGGATAACCTAGGCATACTGCACTG  
TACACTCTGACATATGAAGTGCTCTAGTCAAGTTAACGGTGTCCACAGAGGACAT  
GGTTAACTGGAATTCGTCAAGCCTCTGGTTCTAATTCTCATTGCAG\*GAAATGC  
TGGCATAGAGCAGCACTAAATGACACCACTAAAGAAACGATCAGACAGATCTGGAAT  
GTGAAGCGTTATAGAAGATAACTGGCCTCATTCTCAAAATATCAAGTGTGGAA  
AGAAAAAAAGGAAGTGGAATGGTAACCTTCTGATTAAAAGTTATGTAATAACCAA  
ATGCAATGTGAAATATTTACTGGACTCTTGAAAAACCATCTGAAAAGACTGAG  
GTGGGGGTGGGAGGCCAGCACGGTGGTAGGGCAGTTGAGAAAATTGAATGTGGATT  
AGATTGAAATGATATTGGATAATTATTGGAATTATGGTAATTATGGCCTGTGAGAAGGGTGT  
TGTAGTTATAAAAGACTGTCTTAATTGCATACTTAAGCATTAGGAATGAAGTGT  
TAGAGTGTCTAAATGTTCAAATGGTTAACAAAATGTATGTGAGGCATGTGG  
CAAAATGTTACAGAATCTAACTGGTGGACATGGCTGTCATTGTACTGTTTTCT  
ATCTTCTATATGTTAAAAGTATATAATAAAATATTAATT





**REPLACEMENT SHEET**

**Figure 3A**

1 CGGGGGCCCCACGCTGCGCATCCGGGGTTGCTATGGCGATGAGCAGCAGCAGCAGT  
GGTGGCGGCCTCCGGAGCAGGAGGATTCCGTGCTGTTCCGGCGCCACAGGCCAG  
2 \*AGCGATGATTCTGACATTGGATGATACTGACTGATAAAAGCATATGATAAAGC  
TGTGGCTTCATTAAGCATGCTCTAAAGAACGGTACATTGTGAAACTTCGGGTAA  
ACCAAAAACCACACCTAAAAGAAAACCTGCTAAGAAGAATAAAAGCCAAAAGAAGAA  
3 TACTGCAGCTCCTTACAACAG\*TGGAAAGTTGGGACAAATGTTCTGCCATTGGT  
CAGAAGACGGTTGCATTACCCAGCTACCATTGCTCAATTGATTAAAGAGAGAAA  
CCTGTGTTGGTTACACTGGATATGAAATAGAGAGGAGCAAATCTGCCGATC  
4 TACTTCCCCAATCTGTGAAGTAGCTAATAATATAGAACAGAACGATGCTCAAGAG\*AAT  
GAAAATGAAAGCCAAGTTCAACAGATGAAAGTGAGAACTCCAGGTCTCCTGGAAAT  
AAATCAGATAACATCAAGCCAAATCTGCTCCATGAACTCTTCTCCCTCCACCA  
5 CCCCCCATGCCAGGGCCAAGACTGGGACCAGGAAAG\*CCAGGTCTAAAATTCAATGG  
CCCACCACCGCCACCGCCACCACCAACCCCCACTTACTATCATGCTGGCTGCCTCC  
6 ATTTCTTCTGGACCACCA\*ATAATTCCCCACCACTCCATATGTCCAGATTCTC  
TTGATGATGCTGATGCTTGGAAAGTATGTTAATTGATGGTACATGAGTGGCTATC  
7 ATACTGGCTATTATATG\*GGTTCAAGACAAAATCAAAAAGAAGGAAGGTGCTCACAT  
8 TCCTTAAATTAAAGGA\*GAAATGCTGGCATAGAGCAGCAGCACTAAATGACACCAACTAAAG  
AAACGATCAGACAGATCTGGAATGTGAAGCGTTATAGAACATAACTGGCCTCATTTC  
TTCAAAATATCAAGTGTGGAAAGAAAAAGGAAGTGGAAATGGTAACCTTCTTG  
ATTAAAAGTTATGTAATAACCAATGCAATGTGAAATATTACTGGACTCTTGA  
AAAACCATCTGTAAAAGACTGGGGTGGGGTGGGAGGCCAGCACGGTGGTGGCAG  
TTGAGAAAATTGAATGTGGATTAGATTGAAATGATATTGATAATTATTGTAAT  
TTTATGGCCTGTGAGAAGGGTGTGAGTTATAAAAGACTGTCTTAATTGCATAC  
TTAACGCATTAGGAATGAAGTGTAGAGTGTCTTAAATGTTCAAATGGTTAACAA  
AAATGTATGTGAGGCCTATGTGGCAAAATGTTACAGAACATCTAAACTGGTGGACATGGC  
TGTTCATGTACTGTTCTATCTTATGTTAAAAGTATATAATAAAAAT  
ATTTAATTGTTAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAA  
AAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAGAAAAAAG

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**Figure 3B**

AATTTTAAATTTTAGAGACAGGGTCTCATTATGTTGCCAGGGTGGTGTCAA  
GCTCCAGGTCTCAAGTGATCCCCCTACCTCCGCCTCCAAAGTTGTGGGATTGTAGG  
CATGAGCCACTGCAAGAAAACCTTAAC TG CAG CCT AATA ATT GT TT CTT GGG ATA  
ACTTTAAAGTACATTAAAAGACTATCAACTTAATTCTGATCATATTGTGAAT  
AAAATAAGTAAAATGTCTTGTGAACAAAATGCTTTAACATCCATATAAAGCTATC  
TATATATAGCTATCTATGTCTATATAGCTATTTCACATTCCCTTATTTCCT  
TACAG\*GGTTTCAGACAAAATCAAAAAGAAGGAAGGTGCTCACATTCTAAATTAA  
GGA\*GTAAGTCTGCCAGCATTATGAAAGTGAATCTTACTTTGTAAAACATTATGGT  
TTGTGGAAAACAAATGTTTGAACAGTTAAAAGTTCAAGATGTTAAAAGTTGAAA  
GGTTAATGTAAAACAATCAATATTAAAGAATTGATGCCAAAACATTAGATAAAA  
GGTTAATCTACATCCCTACTAGAATTCTCATACTTAACGGTGGTTATGTGGAAGA  
AACATACTTCACAATAAAGAGCTTAGGATATGATGCCATTATACACTAGTAG  
GCAGACCAGCAGACTTTTTATTGTGATATGGGATAACCTAGGCATACTGCACTG  
TACACTCTGACATATGAAGTGCTCTAGTCAAGTTAACTGGTGTCCACAGAGGACAT  
GGTTAACTGGAATT CGT CAAGC CTG GTT CTA ATT CTC ATT GCAG \*GAAATGC  
TGGCATAGAGCAGCACTAAATGACACCACTAAAGAAACGATCAGACAGATCTGGAAT  
GTGAAGCGTTATAGAAGATAACTGGCCTCATTCTCAAATATCAAGTGTGGAA  
AGAAAAAAAGGAAGTGGAAATGGGTAACTCTTCTGATTAAAAGTTATGTAATAACCAA  
ATGCAATGTGAAATATTTACTGGACTCTTGAAAAACCATCTGTAAGACTGGG  
GTGGGGGTGGGAGGCCAGCACGGTGGTGGCAGTTGAGAAAATTGAATGTGGATT  
AGATTGAAATGATATTGGATAATTATTGGTAATTGATGGCTGTGAGAAGGGTGT  
TGTAGTTATAAAAGACTGTCTTAATTGCATACTTAAGCATTAGGAATGAAGTGT  
TAGAGTGTCTAAATGTTCAAATGGTTAACAAAATGTATGTGAGGCGTATGTGG  
CAAAATGTTACAGAATCTAAGTGGACATGGCTGTCATTGTACTGTTTTCT  
ATCTTCTATATGTTAAAAGTATATAATAAAAATATTAAATT

**Figure 4****C212**

ACCTGANCCCAGANGTCAAGGCTGCAGTGAGACGAGATTGCNCCACTGCCCTCAC  
 CCTGGGTGATAAGAGTGGGACCCTGNTAAAACATACACACACACACACACA  
 CACACACACACACACACACACTCTCTCTCTCTCTCTCTCTCTCTCTCTC  
 TCTCTCTCAAAAACACTGGTCTGTTATTTNCGAAATTGTCAGTCATAGTTATCT  
 GTTAGACCAAAGCTGN GTAAGNACATTATTACATTGCCTCACAACATTCA  
 TAATGTATTTGCTATATAGCAATTACATATNGGNATATATTATCTNAGGGGATGGC  
 CANGTNATAAAACTGTCACTGAGGAAAGGA

**C272**

CCTCCCACCTNAGCCTCCCCAGTAGCTAGGACTATAGCGTGCNCCACCAAGCTCAG  
 CTATTNTNTATTAGTAGAGACGGGTTTCCGGCANGCTAGGCCTCGTNTCGAAC  
 TCCAGTGA  
 GATATTATTCCCCCTCCCCCTGGAAAAGTAAGTAAGCTCCTACTAGGAATTAAA  
 ACCTGCTTGATCTATATAAGACAAACAAGGAAAGACAAACATGGGGCAGGAAGGA  
 AGGCAGATC

**AFM157xd10**

TCGAGGTAGATTGTATTATATCCATGTACACACACACACACACACACACACAC  
 ACACACACACAGACTTAATCTGTTACAGAAATAAAAGGAATAAAATACCGTTCTA  
 CTATACACCAAAACTAGCCATCTTGAC

**C161**

CCCTGAGAAGGCTCCTGAGTATGCATAAACATTACAGCTTGCATGCGTGTGT  
 GTGTGTGTGTGTGTATGTTGCTTGCAGTGTAAAAACAATTGCAACATCAACA  
 GAAATAAAATTAAAGGAATAATTCTCCTCCGACTCTGCCGTTCCATCCAGTGAAC  
 TCTTCATTCTGGGGTAAAGTTCCTTCAGTTCTTCATAGATAGGTATATACTTCATA  
 AGTCAAACAATCAGGCTGGGTGCAGTAGCTCATGCCTGTAATCCCAGCCCTTGGGA  
 GGCGAGCTGGGCAGATCGA

**C171**

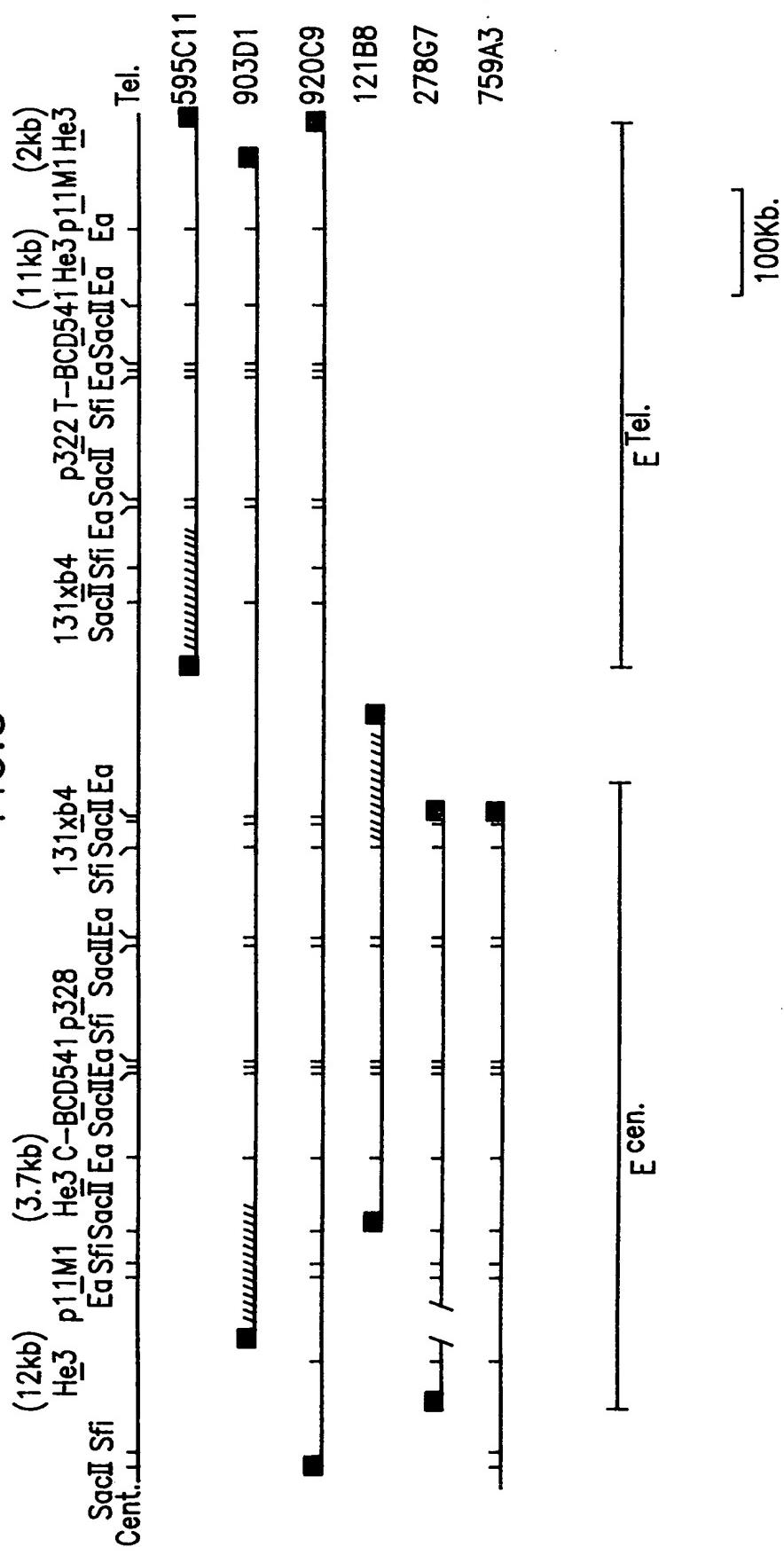
TCCACCCGCCTGGCCTCCAAAGCNCTGGGATTACAGGCGTGACTGCCGCACCCAG  
 CTGTAAACTGGNTNTAAATGGTAGATTTNAGGTATTAACAATAGATAAAAAGATA  
 CTTTNGGCATACTGTGTATTGGGATGGGTTAGAACAGGTGTNCTACCCAAGACAT  
 TTACTTAAATGCCCTCGAAATGCTATGTGAGCTGTGTGTGTGTGTGTGT  
 GTGTATTAAGGAAAGCATGAAAGTATTTATGCTTGTGATTTTTTTNACTCATAG  
 CTTCATAGTGGANCAGATACTAGTCTAAATGTTAAACTTTTATGTCAC  
 TTGCTGTC



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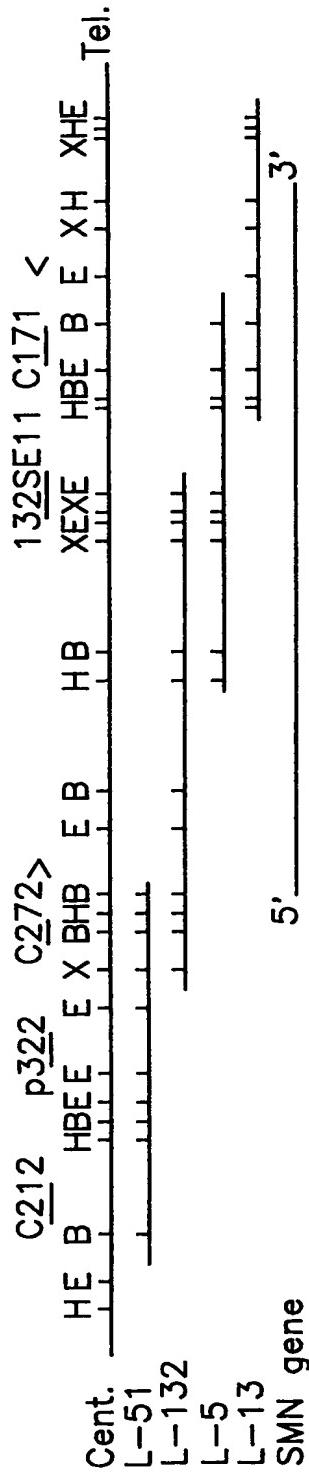
**REPLACEMENT SHEET**

5  
E/G



Restriction map of the 5q13 region for EagI(Ea), SacII(SacII), Sfi(Sfi).Numbers under parenthesis indicate the restriction fragment detected by He3; Telomeric element (E Tel ),centromeric element (E cen ), Centromer(Cent.),Telomere(Tel.).Probes are indicated above the restriction map.YACS are below the restriction map.

FIG.6



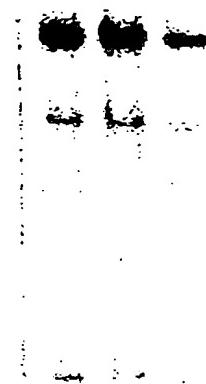
Telomeric element (E Tel) containing the survival motor-neuron gene (SMN gene). Genetic map shows polymorphic markers C212, C272 and C171. Physical map shows location and direction of transcription of SMN gene; phage clones used for assembling physical map. Restriction map for EcoRI(E), XbaI(X), HindIII(H), BglII(B), SacII(S) are shown. Cent. and Tel. indicate centromere and telomere respectively. The position of genomic rearrangements found in SMA patients are also indicated.



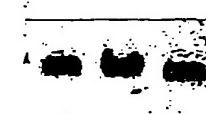


F M A

GENE DOSAGE ANALYSIS OF THE 5q13 REGION WITH THE 132SE11 PLASMID CONE IN SMA TYPE I PATIENT. TOTAL HUMAN DNA FROM SMA FAMILY WAS DIGESTED WITH HindIII FOR SOUTHERN BLOTTING. FILTER WAS CONSECUTIVELY HYBRIDIZED WITH 132SE11 (A) AND JK53 PROBES (B). A SIGNIFICANT DECREASE IN 132SE11 BAND INTENSITY, WHICH INDICATED THE DELETION, COMPARED WITH THEIR PARENTS. F/FATHER, M/MOTHER, A/AFFECTED



A



B

**FIG. 7**



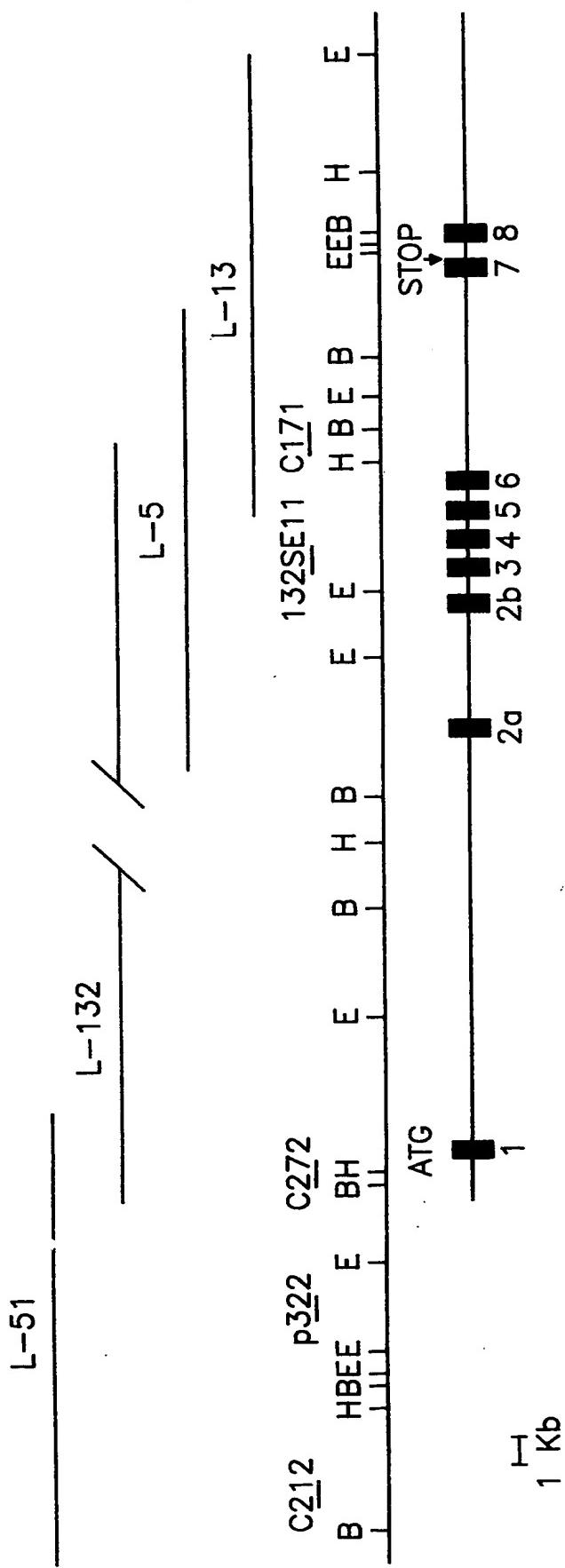
REPLACEMENT SHEET

**Figure 8**

MAMSSGGSGGGVPEQEDSVLFRRGTQSDDSDIWDDTALIKAYDKAVASFKHALKNG  
DICETSGKPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDCIYPATIA  
SIDFKRETCVVVYTGYGNREEQNLSDLSPICEVANNIEQNAQENENESQVSTDESE  
NSRSPGNKSDNIKPKSAPWNSFLPPPPMPGPRLGPGKPGKGLKFNGPPPPPPPHL  
LSCWLPPFPGPPIIPPPPICPDSDLDDADALGSMLISWYMSGYHTGYYM



FIG.9





REPLACEMENT SHEET

**Figure 10A**

**1**

cctccgggaccgtactgttccgtcccagaagccccgggcgccggaagtcgta  
tcttaagaaggacggggccccacgctgcgcacccgcggggttgc ATG GCG

M A

ATG AGC AGC GGC GGC AGT GGT GGC GGC GTC CCG GAG CAG GAG  
M S S G G S G G V P E Q E  
GAT TCC GTG CTG TTC CGG CGC GGC ACA GGC CAG gtgaggtcgac  
D S V L F R R G T G Q

cagtgcagtctccatttagcgcttcagcacccttcttccggccaactctccttc  
cgca

**2a**

gtgtaatttttatgtgtggattaagatgactcttggtaactaacatacatttctg  
attaaacctatctgnacatgagtttttatttacccttccag AGC GAT

S D

GAT TCT GAC ATT TGG GAT GAT ACA GCA CTG ATA AAA GCA TAT  
D S D I W D D T A L I K A Y

GAT AAA GCT GTG GCT TCA TTT AAG gtatgaaatgcttgnttagtcgtt  
D K A V A S F K

tcttattttctcgatttcatttgaaaaggattgataacatacgataaaagtgttaa

**2b**

aggcgcttctgaggtgacggagcctttagacttagttatgtactgggttat  
gtcgtagtttattctgtgcaccaccctgttaacatgtacattttattcctatttt  
cgtag CAT GCT CTA AAG AAT GGT GAC ATT TGT GAA ACT TCG GGT

H A L K N G D I C E T S G

AAA CCA AAA ACC ACA CCT AAA AGA AAA CCT GCT AAG AAG AAT  
K P K T T P K R K P A K K N

AAA AGC CAA AAG AAG AAT ACT GCA GCT TCC TTA CAA CAG gttat  
K S Q K K N T A A S L Q Q

tttaaaatgtgaggatttaacttcaaaggatgtctcattagtccttatttaatagt  
gtaaaatgtctttaact

**3**

gcctgcaggcgatcaaaacgagatgatgtttgcctcttcaaaagaaatgtgtgc  
atgtatatatcttgatttcttttag TGG AAA GTT GGG GAC AAA TGT

W K V G D K C

TCT GCC ATT TGG TCA GAA GAC GGT TGC ATT TAC CCA GCT ACC  
S A I W S E D G C I Y P A T

ATT GCT TCA ATT GAT TTT AAG AGA GAA ACC TGT GTT GTG GTT  
I A S I D F K R E T C V V V

TAC ACT GGA TAT GGA AAT AGA GAG GAG CAA AAT CTG TCC GAT  
Y T G Y G N R E E Q N L S D

CTA CTT TCC CCA ATC TGT GAA GTA GCT AAT AAT ATA GAA CAG  
L L S P I C E V A N N I E Q



**REPLACEMENT SHEET**

**Figure 10B**

AAT GCT CAA GAG gtaaggataaaaaaaaaaaaaattcaatttctggaaagcag  
N A Q E  
agactagatgagaaactgttaaacagtatacaca  
**4**  
ccaccgaggcattaattttcttaatcacacccttataacaaaaacctgcataattt  
tttcttttaaag AAT GAA AAT GAA AGC CAA GTT TCA ACA GAT GAA  
N E N E S Q V S T D E  
AGT GAG AAC TCC AGG TCT CCT GGA AAT AAA TCA GAT AAC ATC  
S E N S R S P G N K S D N I  
AAG CCC AAA TCT GCT CCA TGG AAC TCT TTT CTC CCT CCA CCA  
K P K S A P W N S F L P P P  
CCC CCC ATG CCA GGG CCA AGA CTG GGA CCA GGA AAG gtaaacctt  
P P M P G P R L G P G K  
ctatgaaagtttccagaaaatagttaatgtcgacattaacctctgttaact  
aattttagctctccca  
**5**  
caaatatctggtaatttttatcctttggggtagtcctttattcctat  
catattgaaattggtaagttaatttccttgaaatattccttata CCA GGT  
P G  
CTA AAA TTC AAT GGC CCA CCA CCG CCA CCG CCA CCA CCA CCA  
L K F N G P P P P P P P P P P  
CCC CAC TTA CTA TCA TGC TGG CTG CCT CCA TTT CCT TCT GGA  
P H L L S C W L P P F P S G  
CCA CCA gtaagaaaaagagtataggttagatttgcacataatttga  
P P  
taatta  
**6**  
ccagactttactttgttactggatataacaatatctttctgtctccag  
ATA ATT CCC CCA CCA CCT CCC ATA TGT CCA GAT TCT CTT GAT  
I I P P P P I C P D S L D  
GAT GCT GAT GCT TTG GGA AGT ATG TTA ATT TCA TGG TAC ATG  
D A D A L G S M L I S W Y M  
AGT GGC TAT CAT ACT GGC TAT TAT ATG gtaagtaatcactcagcatct  
S G Y H T G Y Y M  
tttcctgacaattttttgttagttatgtgactttgttggtaaatttataaaaact  
acttg  
**7**  
aactgcagcctaataattgtttcttggataactttaaagtacattaaaagact  
atcaacttaatttctgatcatatttgtgaataaaaataagtaaaatgtcttgtaa



## REPLACEMENT SHEET

Figure 10C

→ a

acaaaatgcttttaacatccatataaagctatctatatatagttatgttat

→ T

atagctatttttaacttccttttatttccttacag GGT TTC AGA CAA

G F R Q

AAT CAA AAA GAA GGA AGG TGC TCA CAT TCC TTA AAT taaggaga

N Q K E G R C S H S L N \*

aagtctgccagcattatgaaagtgaatcttactttgtaaaaactttatggttgtgg

→ g

aaaacaaaatgttttgaacagttaaaaagttcagatgttaaaaagttgaaaggtaa  
tgtaaaaacaatcaatattaagaattttgatgccaaaactattagataaaaaggtaa

→ g

tctacatccctactagaattctcatacttaactgggtggtatgtggaagaaacata  
ctttcacaataaagagctttaggatatgatgccatttatatactacttaggcagac  
cagcagacttttttattgtgatatgggataacctaggcatactgcactgtacact  
ctgacatatgaagtgcttagtcaagtttaactgggtccacagaggacatggutta

8

actggaattcgtcaagcctctggttctaatttctcattgcagggaaatgctggcata  
gagcagcactaaatgacaccactaaagaaacgatcagacagatctggaatgtgaagc  
gttatagaagataactggcctcattcttcaaaatatacaagtgttggaaagaaaaa  
aggaagtggaaatggtaactcttcttgattaaaagttatgtaataaccataatgcaat

→ a

gtgaaatatttactggactctttgaaaaaccatctagaaaagactgggggggg  
gtgggaggccagcacggtggtgaggcagttgagaaaaattgaatgtggattagatt  
tgaatgatattggataatttggtaattttatggcctgtgagaagggttgttagt  
ttataaaagactgtcttaatttgcatacttaagcatttaggaatgaagtgttagat  
gtcttaaaatgtttcaaattggtaacaaaatgtatgtgaggcgtatgtggaaaaat  
gttacagaatctaactggtgacatggctgttcatgtactgttttttatcttc  
tatatgtttaaaagtataataaaaaatattta



**REPLACEMENT SHEET**

**Figure 11**



## REPLACEMENT SHEET

Figure 12A

cggcgtggtagcaggcc	ATG GCG ATG GGC AGT GGC GGA GCG	41
	Met Ala Met Gly Ser Gly Gly Ala	
GGC TCC GAG CAG GAA GAT ACG GTG CTG TTC CGG CGT GGC		80
Gly Ser Glu Gln Glu Asp Thr Val Leu Phe Arg Arg Gly		
ACC GGC CAG AGT GAT GAT TCT GAC ATT TGG GAT GAT ACA		119
Thr Gly Gln Ser Asp Asp Ser Asp Ile Trp Asp Asp Thr		
GCA TTG ATA AAA GCT TAT GAT AAA GCT GTG GCT TCC TTT		158
Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala Ser Phe		
AAG CAT GCT CTA AAG AAC GGT GAC ATT TGT GAA ACT CCA		197
Lys His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr Pro		
GAT AAG CCA AAA GGC ACA GCC AGA AGA AAA CCT GCC AAG		236
Asp Lys Pro Lys Gly Thr Ala Arg Arg Lys Pro Ala Lys		
AAG AAT AAA AGC CAA AAG AAG AAT GCC ACA ACT CCC TTG		275
Lys Asn Lys Ser Gln Lys Lys Asn Ala Thr Thr Pro Leu		
AAA CAG TGG AAA GTT GGT GAC AAG TGT TCT GCT GTT TGG		314
Lys Gln Trp Lys Val Gly Asp Lys Cys Ser Ala Val Trp		
TCA GAA GAC GGC TGC ATT TAC CCA GCT ACT ATT ACG TCC		353
Ser Glu Asp Gly Cys Ile Tyr Pro Ala Thr Ile Thr Ser		
ATT GAC TTT AAG AGA GAA ACC TGT GTC GTG GTT TAT ACT		392
Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr Thr		
GGA TAT GGA AAC AGA GAG GAG CAA AAC TTA TCT GAC CTA		431
Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu		
CTT TCC CCG ACC TGT GAA GTA GCT AAT AGT ACA GAA CAG		470
Leu Ser Pro Thr Cys Glu Val Ala Asn Ser Thr Glu Gln		
AAC ACT CAG GAG AAT GAA AGT CAA GTT TCC ACA GAC GAC		509
Asn Thr Gln Glu Asn Glu Ser Gln Val Ser Thr Asp Asp		
AGT GAA CAC TCC TCC AGA TCG CTC AGA AGT AAA GCA CAC		548
Ser Glu His Ser Ser Arg Ser Leu Arg Ser Lys Ala His		



## REPLACEMENT SHEET

Figure 12B

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AGC AAG TCC AAA GCT GCT CCG TGG ACC TCA TTT CTT CCT	587
Ser Lys Ser Lys Ala Ala Pro Trp Thr Ser Phe Leu Pro	
CCA CCA CCC CCA ATG CCA GGG TCA GGA TTA GGA CCA GGA	626
Pro Pro Pro Pro Met Pro Gly Ser Gly Leu Gly Pro Gly	
AAG CCA GGT CTA AAA TTC AAC GGC CCG CCG CCG CCT	665
Lys Pro Gly Leu Lys Phe Asn Gly Pro Pro Pro Pro	
CCA CTA CCC CCT CCC CCC TTC CTG CCG TGC TGG ATG CCC	704
Pro Leu Pro Pro Pro Phe Leu Pro Cys Trp Met Pro	
CCG TTC CCT TCA GGA CCA CCA ATA ATC CCG CCA CCC CCT	743
Pro Phe Pro Ser Gly Pro Pro Ile Ile Pro Pro Pro Pro	
CCC ATC TCT CCC GAC TGT CTG GAT GAC ACT GAT GCC CTG	782
Pro Ile Ser Pro Asp Cys Leu Asp Asp Thr Asp Ala Leu	
GGC AGT ATG CTA ATC TCT TGG TAC ATG AGT GGC TAC CAC	821
Gly Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr His	
ACT GGC TAC TAT ATG GGT TTC AGA CAA AAT AAA AAA GAA	860
Thr Gly Tyr Tyr Met Gly Phe Arg Gln Asn Lys Lys Glu	
GGA AAG TGC TCA CAT ACA AAT taag	885
Gly Lys Cys Ser His Thr Asn *	

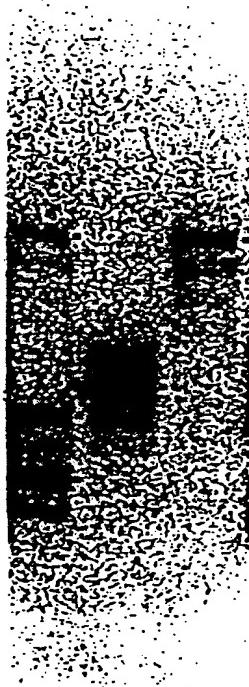


**REPLACEMENT SHEET**

**Figure 13**

20 30 40 50 60 70  
 GSGGGVPEQEDSVLFRRGTGQSDDSDIWDDTALIKAYDKAVASFKHALKNGDICETS  
 =====  
 GSGGAGSEQEDTVLFRRGTGQSDDSDIWDDTALIKAYDKAVASFKHALKNGDICETP  
 20 30 40 50 60  
 80 90 100 110 120 130  
 GKPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPATIASIDFKR  
 =====  
 DKPKGTARRKPAKKNKSQKKNATTPLKQWKVGDKCSAVWSEDGCIYPATITSIDFKR  
 70 90 100 110 120  
 140 150 160 170 180 1  
 ETCVVVYTGYGNREEQNLSDLLSPICEVANNIEQNAQENENESQVSTDESENSRSPG  
 =====  
 ETCVVVYTGYGNREEQNLSDLLSPTCEVANSTEQNTQENE--SQVSTDDSEHSSRSL  
 130 140 160 170 1  
 90 200 210 220 230 240  
 NKSDNIKPKSAPWNSFLPPPPMPGPRLGPGKPGLKFNGLNGPPPPPPPHLLSCWLP  
 =====  
 RSKAHSKSKAAPWTSFLPPPPMPGSGLGPGKPGLKFNGLNGPPPPPLPPPFLLPCWMP  
 80 190 200 210 220 230  
 250 260 270 280 290 300  
 PFPSGPPIIPPPPICPDSDLDDADALGSMLISWYMSGYHTGYMGFRQNQKEGRCSH  
 =====  
 PFPSGPPIIPPPPICPDCLDDTDALGSMLISWYMSGYHTGYMGFRQNQKEGKCSH  
 240 250 260 270 280 290

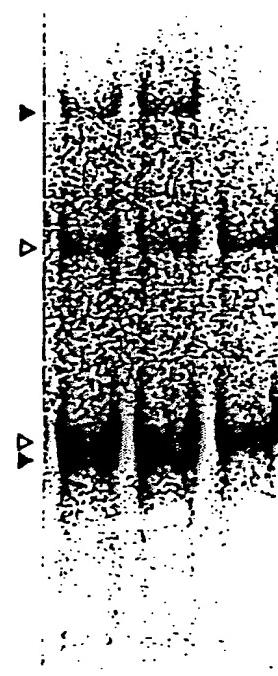
SL  
—  
TN



F M A



F M A



F M A

**FIG. 14(A)**

**FIG. 14(B)**

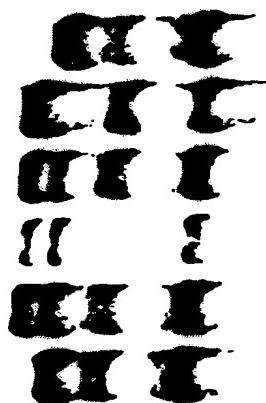
**FIG. 14(C)**



## SSCP ANALYSIS

▼ SMN  
▽ C-BCD541

▼ ▽



121B8 YAC  
595CII YAC  
HUMAN 1 CONTROL  
HUMAN 2 CONTROL  
HUMAN 3 CONTROL  
HUMAN 4 SMA

# FIG. 15

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